IL1A gene

interleukin 1 alpha

Normal Function

The *IL1A* gene provides instructions for making a protein called interleukin-1 alpha. Interleukins are a group of proteins that are made primarily in immune system cells. They are involved in cell-to-cell communication and have a wide variety of functions within the immune system. Interleukin-1 alpha is described as "pro-inflammatory" because it stimulates the activity of genes involved in inflammation and immunity. This protein plays a critical role in protecting the body from foreign invaders such as bacteria and viruses. It is also involved in bone resorption, the breakdown and removal of bone tissue that is no longer needed.

Interleukin-1 alpha is initially produced as a relatively long protein that is trapped within cells. Another protein, called calpain, cuts (cleaves) this precursor protein to create a shorter, mature version of interleukin-1 alpha. The shorter form of this protein is secreted by immune system cells to influence the functions of other cells.

Health Conditions Related to Genetic Changes

ankylosing spondylitis

Several variations (polymorphisms) in the *IL1A* gene have been found to influence the risk of ankylosing spondylitis. Each of these variations changes a single protein building block (amino acid) in interleukin-1 alpha. It is unclear how these variations alter the protein's function. Studies suggest that the effects of *IL1A* variations are probably related to the role of interleukin-1 alpha in promoting inflammation. Other genetic and environmental factors, many of which are unknown, also affect the chance of developing ankylosing spondylitis.

idiopathic inflammatory myopathy

intervertebral disc disease

other disorders

Variations in the *IL1A* gene have been studied as potential risk factors for several other disorders associated with abnormal inflammation. These include chronic gum (periodontal) disease, a progressive bone infection known as chronic osteomyelitis, and an eye disease called open-angle glaucoma.

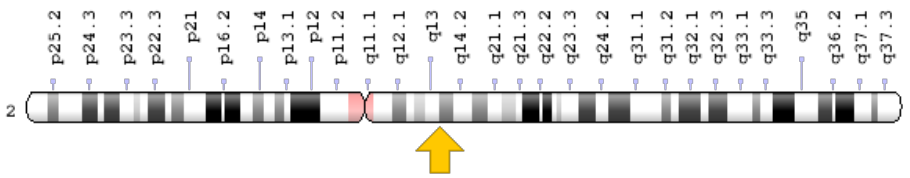
The most well-studied variation affecting the *IL1A* gene is a change in a single DNA building block (nucleotide) in a region of regulatory DNA near the start of the gene (written as IL1A-889 C>T). This variation affects the production of interleukin-1 alpha within cells. Researchers have also identified a second common variation in the *IL1A* gene, written as IL1A+4845 G>T, which changes a single nucleotide in the gene. This variation likely affects the sensitivity of interleukin-1 alpha to cleavage by calpain.

It is unclear how changes in the *IL1A* gene influence the risk of inflammatory disorders. Studies suggest that the effects of *IL1A* variations are probably related to the role of interleukin-1 alpha in promoting inflammation. Other genetic and environmental factors also likely affect the chance of developing these complex disorders.

Chromosomal Location

Cytogenetic Location: 2q14.1, which is the long (q) arm of chromosome 2 at position 14.1

Molecular Location: base pairs 112,773,915 to 112,785,398 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- hematopoietin-1
- IL-1 alpha
- IL-1A
- IL1
- IL1-ALPHA
- IL1A_HUMAN
- IL1F1
- Interleukin-1 alpha
- interleukin 1, alpha

- preinterleukin 1 alpha
- pro-interleukin-1-alpha

Additional Information & Resources

Educational Resources

- National Eye Institute: Glaucoma
  https://nei.nih.gov/health/glaucoma/glaucoma_facts
- National Institute of Arthritis and Musculoskeletal and Skin Diseases: Rheumatoid Arthritis
  https://www.niams.nih.gov/Health_Info/Rheumatic_Disease/

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28IL1A%5BTI%5D%29+OR+%28IL-1+alpha%5BTI%5D%29+OR+%28IL-1A%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- INTERLEUKIN 1-ALPHA
  http://omim.org/entry/147760

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_IL1A.html
- HGNC Gene Family: Endogenous ligands
  http://www.genenames.org/cgi-bin/genefamilies/set/542
- HGNC Gene Family: Interleukins
  http://www.genenames.org/cgi-bin/genefamilies/set/601
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5991
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/3552
- UniProt
  http://www.uniprot.org/uniprot/P01583

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